orphanaesthesia

Anaesthesia recommendations for

Congenital cataracts, facial dysmorphism and neuropathy syndrome

Disease name: Congenital cataracts, facial dysmorphism and neuropathy syndrome

ICD 10: Q87.8

Synonyms: CCFDN syndrome

Disease summary: Congenital cataracts, facial dysmorphism and neuropathy (CCFDN) syndrome is an extremely rare autosomal recessive disorder with unknown prevalence. This multiorgan disorder is typically described in Roma ethnicity. The first case was described in 1999 in Roma patients from Bulgaria. It is caused and diagnosed by a mutation in CTDP1 gene on chromosome 18q23. This mutation causes an altered transcription process, affecting many cellular processes' functions. CCFDN syndrome has a similar clinical manifestation to Marinesco-Sjögren syndrome, but molecular testing showed that these syndromes are different. Disease abnormalities include ophthalmic problems, especially bilateral congenital cataract, nystagmus or microcorneae, facial dysmorphism with micrognathia, mild development delay, musculoskeletal deformities caused by demyelinating peripheral neuropathy and hypogonadism. These patients undergo ophthalmic surgery of cataracts, corrective orthopaedic surgery like scoliosis or extremities correction. Perioperative management includes close anaesthetic monitoring, post-operative care in ICU is appropriate, except for short non-complicated surgery, due to potentially life-threatening complications like epileptic seizures, rhabdomyolysis, pulmonary oedema or inspiratory stridor. The main post-operative complication of patients with CCFDN syndrome is rhabdomyolysis, so we should limit using volatile anaesthetics and depolarising muscle relaxants. Total intravenous anaesthesia, eventually non-depolarising muscle relaxants, are preferred.

Medicine is in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnosis is wrong

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Typical surgery

Most frequent procedures include ophthalmic surgery, typically cataract, orthopaedic surgery like scoliosis or extremities correction and tendon transfers. Rarer procedures are corrections of gonad abnormalities.

Type of anaesthesia

There are possible both kinds of anaesthesia, general or regional anaesthesia. General anaesthesia is the preferred method for a patient suffering from mental retardation and bad compliance.

There are unknown specific contraindications for regional anaesthesia associated with CCFDN syndrome, but there is just one published report about regional anaesthesia in these patients. Ultrasound navigated peripheral blocks are useful as part of postoperative analgesia, but cannulation can be difficult due to different anatomical proportions in patients with musculoskeletal deformations.

General anaesthesia is a method of choice in patients with altered mental status. Depolarising muscle relaxants and volatile anaesthetics should be limited. There are described high risk of severe rhabdomyolysis and hyperkalaemia. However, rhabdomyolysis is probably triggered by stress and ineffective postoperative pain control. The inhalational induction to the anaesthesia can be considered in case of difficult IV access to reduce the stress. Total intravenous anaesthesia, eventually with non-depolarising muscle relaxants in case of intubation or to facilitate the surgery, are the best choice. Rocuronium and sugammadex administration is preferred combination to neuromuscular blockade control, there is a potential to full recovery from the neuromuscular blockade. There are unconvincing data about malignant hyperthermia, but the association with malignant hyperthermia is unlikely due to the different gene localisation of these disorders.

Analgosedation is not contraindicated, but it should be considered individually, especially in patients with altered respiratory functions due to scoliosis, mental retardation or muscle weakness. In addition, facial dysmorphism, specifically mandibular hypoplasia can cause upper airway obstruction after sedation.

Necessary additional pre-operative testing (beside standard care)

The spectrum of pre-operative assessment should be considered individually according to the severity of symptoms and the type of surgery. CCFDN syndrome is characterised by demyelinating peripheral neuropathy with skeletal muscle weakness. It can result in scoliosis with cardiopulmonary functions alteration. Pre-anaesthesia evaluation should focus on the signs of difficult airway management in the first place and on the identification of cardiovascular and pulmonary disorders and neurological deficits, respectively muscle weakness.

Cardiovascular assessment includes electrocardiography. There are no data about serious cardiac disease, but in case of doubts (severe scoliosis), echocardiography should be considered.

Respiratory function evaluation should be considered in patients with severe muscle weakness or severe scoliosis. It includes arterial blood gas analysis and spirometry. This testing can be

complicated by the non-compliant patient with mental retardation, but it can be helpful for the prediction of the need for post-operative ventilatory support.

Neurological examination is recommended in patients with neurological deficit, severe muscle weakness, extremity deformations and before spine surgery. In addition, it can be helpful for juridical reasons in these patients for a clear description of neurological deficit in case of possible iatrogenic harm of patients.

Particular preparation for airway management

CCFDN syndrome is a multiorgan disorder. Facial dysmorphism is a typical sign of this syndrome, especially in late childhood or adult males. Dysmorphism is presented by prominent nasal philtrum with upper incisors, thickening of the perioral tissues and micrognathia. Two cases report different types of airway securement. A laryngeal mask was used in the first published case, endotracheal intubation in the second one. Both cases do not report problems with bag-mask ventilation or airway securing.

There are no more data about airway securing, for the possibility of difficult airways due to facial dysmorphism, the equipment for difficult airway management should be available before every anaesthetic care.

Particular preparation for transfusion or administration of blood products

Data about transfusion management have not been published. There is expected higher blood loss and need for blood products in a patient with CCFDN syndrome during invasive procedures like spine surgery or extremities correction. Some data presented higher blood loss in patients with the neuromuscular disorder compared to patients without neuromuscular deficit. There is probably the role of osteoporosis for higher blood loss in these patients.

Particular preparation for anticoagulation

There is no case about thrombotic complications in the literature. Recommendation for anticoagulation therapy is not available. But there is the expectation of a higher risk for thrombosis in patients with limited mobility. We should consider the risk vs benefit from anticoagulation in the perioperative period in these patients.

Particular precautions for positioning, transportation and mobilisation

Patients with CCFDN syndrome can suffer from osteoporosis due to peripheral neuropathy with low mobility level or endocrine deficiency. There is a higher risk of iatrogenic injury in these patients. It is recommended to use specific positioning pads in these patients, especially in the prone position.

Interactions of chronic disease and anaesthesia medications

Not reported.

Anaesthetic procedure

Regional anaesthesia, neuraxial techniques including, are possible with respect to the mental status of these patients. However, regional techniques can be combined with general anaesthesia. There is no specific approach, but there could be different anatomical proportions in patients with neuromuscular disease. Ultrasound is the preferred method for regional anaesthesia navigation.

General anaesthesia is preferred in a patient with severe mental alteration. Total intravenous anaesthesia is a method of choice.

Volatile anaesthetics and depolarising muscle relaxants should be limited. There is a higher risk of hyperkalaemia and rhabdomyolysis.

Non-depolarising muscle relaxants can be used in patients with CCFDN syndrome. We recommend neuromuscular blockade monitoring due to the higher risk of prolonged neuromuscular blockade.

Good pain control prevents the risk of excessive stress reaction and rhabdomyolysis. Regional anaesthesia and paracetamol were used without complications.

Particular or additional monitoring

Depth of anaesthesia should always be monitored during TIVA. It can reduce the total dose of anaesthetics, and it shortens the time of recovery from anaesthesia.

We should always monitor the depth of neuromuscular blockade. There is a risk of prolonged neuromuscular blockade in patients with demyelinating peripheral neuropathy.

Invasive blood pressure monitoring is recommended in more extensive surgery, major fluid shifts or in patients with altered cardiovascular functions.

Possible complications

Patients with CCFDN syndrome are at higher risk of rhabdomyolysis. Volatile anaesthetics should be limited. The potential rhabdomyolysis can be detected postoperatively by myoglobinuria monitoring.

There are described other complications like seizures, pulmonary oedema or inspiratory stridor. These complications should be treated standardly.

Peripheral neuropathy should be associated with prolonged neuromuscular blockade. There is a risk of impaired recovery due to residual neuromuscular blockade.

Post-operative care

The post-operative monitoring depends on the type of surgical procedure and patient comorbidities. Intensive care should be provided in high-risk surgeries in patients with high degree disability.

The potential rhabdomyolysis can be detected postoperatively by myoglobinuria monitoring.

Patients could profit from early weaning and mobilisation after surgery. Immobilisation can worsen the neuropathy and the outcome after the surgery.

Disease-related acute problems and effect on anaesthesia and recovery

There are limited data about airway management. Difficult airways should be expected always due to facial dysmorphism.

There are unconvincing data about malignant hyperthermia. But the association with malignant hyperthermia is unlikely because of the different gene localisation of CCFDN syndrome and malignant hyperthermia.

Ambulatory anaesthesia

Not reported.

Obstetrical anaesthesia

Not reported.

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Please note that this recommendation has not been by an anaesthesiologist and a disease expert but by two anaesthesiologists instead.